A “How to” Approach: Genetics, Genomics and Pharmacogenomics for Advanced Practice Nursing Education

Jean Boucher, PhD, ANP-BC & Omanand Koul, PhD, Graduate School of Nursing, University of Massachusetts Worcester
Widening gap and growing demands with advances in genetics, genomics and pharmacogenomics

- Outpacing the knowledge base in advanced practice nurses prepared to provide care in acute, urban or rural community settings

- Personalized medicine, predictive and direct-to-consumer genetic testing, and use of targeted therapies.

- Genetics and genomics healthcare content includes *Essential Nursing Competencies and Curricular Guidelines for Genetics and Genomics in AACN Essentials*
Objectives related to learning needs:

- Basic principles of human genetics, cytogenetics, heredity, and genomics.
- Clinical applications of genetics, genomics and molecular and cellular biology.
- Genetic at-risk assessment, referral for genetic counseling & testing.
- Pharmacogenomics in different therapeutic areas.
- Current and future literature in the area of pharmacogenomics.
- Societal and ethical implications (ELSI) of genetic testing and the resultant individualization of drug therapy.
- Disease-related conditions: Alzheimer’s disease, Down’s syndrome, Cystic fibrosis, Cancer, ALS, Huntington’s chorea...
Innovative methodology of hybrid courses

- Classical genetics
- Molecular genetics
- Cytogenetics
- Population genomics
- Pharmacogenomics

- Molecular genetics & cytogenetics
- Newborn screening personalized medicine.
- Genomics, chromosomal, mitochondrial and impact of genomics on society.
- Pedigrees and pedigree analysis in family history
- GWAS (Genetic-wide association studies) and personalized care
- Pharmacogenomics of drug transporters, drug targets, adverse reactions, receptors, therapies, diseases
- Case studies & “telling stories”
Non disjunction:
Autosomal Dominant & Recessive
X linked Dominant & Recessive
Pedigree Analysis

How to Read Pedigrees

- □ = male
- ○ = female
- ♠ or ♦ = homozygous individual who shows the trait (here, albinos)
- ♠ or ♦ = heterozygous carrier of the trait
- ± = offspring
- I, II, III, IV, or V = generation
Inheriting genetic factors increases your risk of developing the disease, but does not make it inevitable.

Knowing which diseases run in your family can help predict your personal risk and help you possibly prevent developing the diseases.
What's Your Family Health History?

What information is important to collect for each family member?

- Gender
- Date of birth
- For deceased relatives, age at the time of death and cause of death
- Diseases or other medical conditions
- Age of disease onset
- Diet, exercise habits, smoking habits or history of weight problems
- Ancestry
Population Genomics: Genome Analysis

- Protein structure analysis and interactions
- Protein sequence analysis
- Epigenetics & Epigenomics
- Studying genetic variations
- Linkage analysis and complex traits
- Application of principles in population genetics
Pharmacogenomics

Pharmacogenomics in the clinic. Health professionals take blood samples from patients with the same condition. DNA is purified from the blood and placed on a profiling chip. The chip tests for gene variants that affect response to a drug used to treat the condition. Depending on which genetic variants they have, patients may have a good response, no response or bad side effects. The drug is given only to people who are likely to have a good response.

Source: National Institutes of Health
http://www.genome.gov/images/illustrations/Pharmacogenomics.pdf
Methodologies

- Group work: Disease related presentations
- Web-based virtual labs: Live and virtual “patient as teachers”: Telling Stories & Genomics Care Webinars (JNS)
- Exercises: Non-Disjunction, Karyotypes, DNA, color blindness, Hardy Weinberg Population Genomics
- Interprofessional collaboration: Genetics counselor lecture
Genomics tv lectures:

- **Genomics introduction:**
  [http://www.youtube.com/watch?v=N4i6lYfYQzY](http://www.youtube.com/watch?v=N4i6lYfYQzY)

- **What is genomics: Lecture by Francis Collins**
  [http://www.youtube.com/watch?v=RvNXPC7qOxY#t=12m02s](http://www.youtube.com/watch?v=RvNXPC7qOxY#t=12m02s)

- **Genome wide association studies**
  [http://www.youtube.com/watch?v=b9GIBWqpinE](http://www.youtube.com/watch?v=b9GIBWqpinE)
Listen to patient stories: Knowledge and competencies

• Bowel cancer:
  Paul’s story: Genetics knowledge for practice
  http://www.tellingstories.nhs.uk/stories.asp?id=38

• Lowri story: Hypercholesterolemia: healthcare provider story: Communication

Patients as Teachers

• Patients “as teachers” come to speak to the class about their experience with being diagnosed with an inherited genetic disease.
Outcomes

• Appropriate risk assessment
• Genetic testing and counseling referrals
• Follow-up preventative care and early detection including patient/family resources
• Ethical, legal, and social implications (ELSI)
• Health disparities, financial aspects and psychosocial supportive needs for at-risk patients and their families.
• Understanding mechanisms of target therapies/pharmacogenomics
• Personalized medicine continues to evolve!
Resources:

  www.NHGRI.org (National Human Genome Research Institute National Institutes of Health)


- National Coalition for Health Professional Education in Genetics (NCHPEG): http://www.nchpeg.org

- National Institutes of Health: http://www.nih.gov

- Healthy People 2020: http://www.healthypeople.gov

- Genome.gov

- UK National Health Service Genetics Education Resource: http://www.tellinestories.nhs.uk/stories


• The *Positives*...online quizzes, assignments, & lectures but also in-class discussion and clarification of materials needed.

• The *Challenges*: Lots of information, knowledge & skills, new discoveries which involves lifelong learning.
  Health disparities, access to care, linguistics, and follow-up care.

• The *Opportunities* to consider genetics/genomics healthcare roles and genetics research opportunities (JNS blueprint article)
How well are we prepared?  Questions?